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Increased nuchal translucency and diaphragmatic hernia. A case report

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Summary

Increased nuchal translucency (NT) thickness is present in 40% of fetuses with diaphragmatic hernia, including 80% of those that result in neonatal death and in 20% of the survivors. A 33-year-old nulliparous woman had first trimester scan at 12 weeks. The fetus had a NT of 2.3 mm, normal ductus venosus (DV), and tricuspid doppler and present nasal bone. Pregnancy-associated plasma protein A (PAPP-A) was 0.59 MoM and beta-human chorionic gonadotropin (b-hCG) 2.56 MoM. The couple did not opt for chorionic villous sampling (CVS) and repeat ultrasound examination was advised. At 18 weeks, ultrasound revealed left sided diaphragmatic hernia. The couple consented for termination of the pregnancy. The molecular test showed normal karyotype and male gender. In such cases with intrathoracic herniation of abdominal viscera, the increased NT may be the consequence of venous congestion due to mediastinal compression. The prolonged compression of the lungs causes pulmonary hypoplasia. Increased NT with normal fetal karyotype is associated with structural fetal anomalies like diaphragmatic hernia and screening at 16-18 weeks is imperative.

Key words: Nuchal translucency; Diaphragmatic hernia; Congenital diseases; Fetal ultrasound scan; Serum screening.

Introduction

Congenital diaphragmatic hernia occurs in approximately one in 4,000 births. Development of the diaphragm is usually completed by the ninth week of gestation. In the presence of the defective diaphragm, there is a herniation of the abdominal viscera into the thorax at about ten to 12 weeks of gestation, when the intestines return to the abdominal cavity from the umbilical cord. However, at least in some cases, intrathoracic herniation of viscera may be delayed until the second or third trimester of pregnancy. When diaphragmatic hernia is present, there is reduction of the available thoracic space to the developing lungs and thus, reduction of the airways, alveoli, and arteries. Furthermore as a consequence, there is an increase in arterial medial wall thickness, and there is also an extension of muscle distally into the small pre-acinar arteries. Although an isolated diaphragmatic hernia is an anatomically simple defect, which might be correctable, the mortality rate is about 50%. The main cause of death is hypoxemia due to pulmonary hypertension, resulting from the abnormal development of the pulmonary vascular bed. In fetuses with a diaphragmatic defect, which allows the intrathoracic herniation of abdominal viscera only after mid-gestation, prenatal correction, by allowing further development of the alveoli and intra-acinar vessels, may well prevent pulmonary hypoplasia and neonatal death [1-4].

The authors present an interesting case of a fetus with increased nuchal translucency (NT), normal karyotype, and left-sided diaphragmatic hernia diagnosed by ultrasound at 18 weeks of gestation. They also present a brief review of the literature about occurrence, follow up, and diagnosis of increased NT normal karyotype, and isolated structural fetal abnormalities.

Case Report

A 32-year-old nulliparous pregnant woman attended the present outpatient department for her routine 1st trimester ultrasound screening for Down's syndrome. On ultrasound assessment the fetus was found to have a NT of 2.3 mm, normal ductus venosus (DV) doppler and present nasal bone (Figure 1). No anatomical abnormalities were identified at the time of the examination. Pregnancy associated placenta protein A (PAPP-A) was 0.59 MoM and beta-human chorionic gonadotropin (b-hCG) 2.56 MoM, whereas the total risk for trisomy 21 was calculated 1/269, by using the Astraia risk calculation system. The couple was offered but did not opt for chorionic villous sampling (CVS). Thus, a repeat ultrasound examination was advised. At 18 weeks, she had a repeat ultrasound which revealed a left-sided diaphragmatic hernia, with prolapsing stomach, and bowel into the thoracic cavity, pushing the heart and the mediastinum towards the right side (Figures 2, 3).

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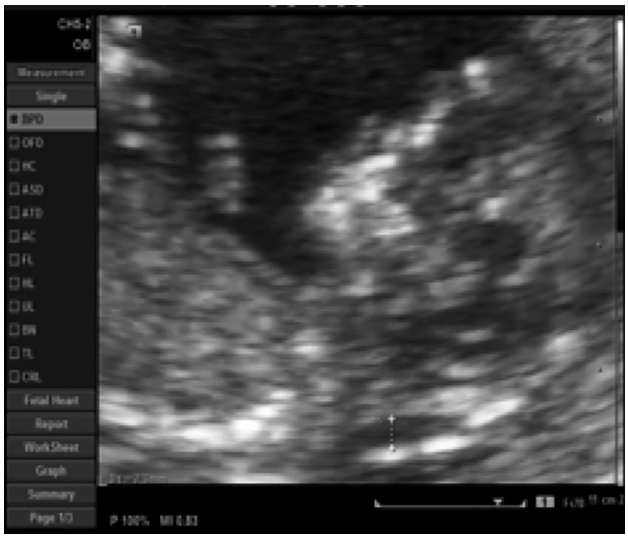

Figure 1. — Ultrasound image of the fetus at 12 weeks, showing the nuchal translucency and present nasal bone.

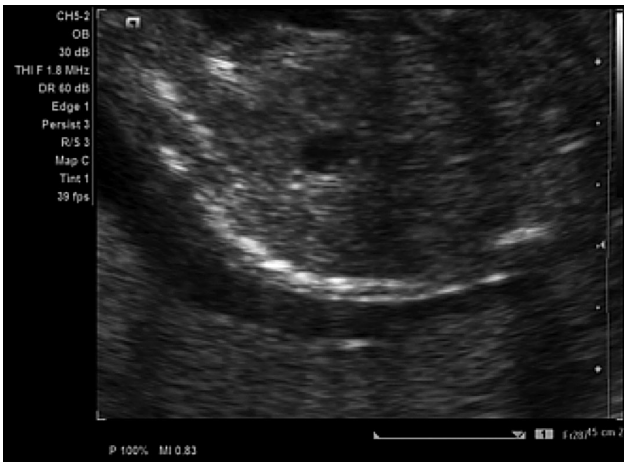

Figure 2. — Ultrasound image showing the diaphragmatic hernia at 18 weeks (sagital view).

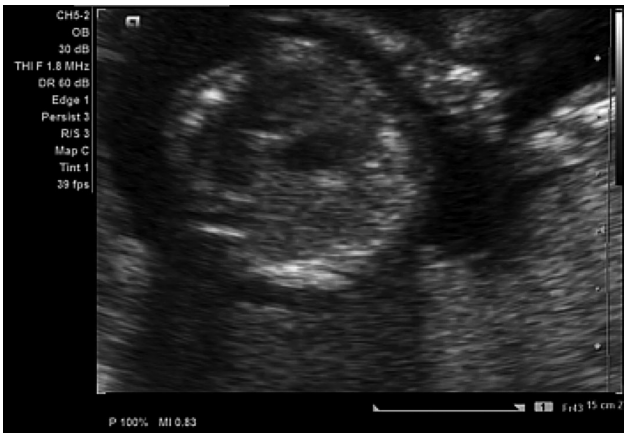

Figure 3. — Ultrasound image showing the diaphragmatic hernia at 18 weeks (transverse view).

Results

The couple was counseled about the poor prognosis of the fetus and they were offered a medical termination. The molecular test of fetal tissue showed a normal karyotype and male gender.

Discussion

Diaphragmatic hernia is usually a sporadic abnormality. However, in about 50% of affected fetuses, there are associated chromosomal abnormalities – usually trisomy 18, 13 – mainly craniospinal defects, including spina bifida, hydrocephalus, and rarely iniencephaly, cardiac abnormalities, and some genetic syndromes such as Marfan, Fryns, and De Lange. If a large defect is present, the fetus may suffer from a severe cardiac failure which leads to ascites, accompanied by severe pulmonary hypoplasia and respiratory insufficiency, causing death in newborns. Mortality rates of the sole cardiac failure are about 60-80%, whereas if accompanied by pulmonary hypoplasia, the rates rise up to 100%. The earlier in gestation the abnormality is present, the poorer the outcome for the fetus [5-8].

Diaphragmatic hernia can be diagnosed by demonstrating the stomach and intestines in 95%, or the liver in 50% of cases protruding in the thorax, and by the mediastinal shift to the opposite side. Herniated abdominal contents associated with a left sided diaphragmatic hernia, are usually easy to demonstrate because of the characteristic imaging of the echo-free fluid filled stomach and the small bowel contrast to the a more echogenic fetal lung. On the contrary, the diagnosis of a right-sided hernia is more demanding, since the echogenicity of the fetal liver is similar to that of the lung, and the visualization of the gall bladder in the right side of the fetal chest, may be the only marker which can lead to the diagnosis. Polyhydramnios, is usually found only after 25 weeks of gestation, in about 75% of the cases, as a consequence of impaired fetal swallowing due to compression of the esophagus by the herniated abdominal organs. If much of the bowel is in the fetal chest, this can result in a reduced abdominal circumference measurement, giving the impression of asymmetrical growth restriction. However, serial measurements, usually demonstrate normal growth velocity. Diagnosis at 18 weeks as in the present case in quiet demanding and difficult since the herniated mass is usually small and there are no clinical suspicious signs, like polyhydramnios at this stage of pregnancy [9-12].

When there is increased NT but the karyotype is normal, the pregnancy remains as high risk for further non chromosomal anomalies and a detailed ultrasound at 18 and 22 weeks should be offered in these cases [2,6,11].

Conclusion

Increased NT with normal fetal karyotype is associated with increased structural fetal anomalies like diaphragmatic hernia and screening for these at 16-18 weeks is imperative.

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